Overview

Useful For

Obtaining a rapid, expert opinion on muscle biopsy specimens for diagnosis of acquired or inherited muscle diseases

Guiding treatment and genetic testing, as well as investigating relevance of genetic variants of unknown significance

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
IHPCI	IHC Initial	No, (Bill Only)	No
IHPCA	IHC Additional	No, (Bill Only)	No
IFPCI	IF Initial	No, (Bill Only)	No
IFPCA	IF Additional	No, (Bill Only)	No
SS2PC	SpecStain, Grp II, other	No, (Bill Only)	No
SS3PC	SpecStain, Grp III, enzyme	No, (Bill Only)	No
HCFPC	SpecStain, frozen	No, (Bill Only)	No
COSPC	Consult, Outside Slide	No, (Bill Only)	No
CSPPC	Consult, w/Slide Prep	No, (Bill Only)	No
CUPPC	Consult, w/USS Prof	No, (Bill Only)	No
CRHPC	Consult, w/Comp Rvw of His	No, (Bill Only)	No
LV4RP	Level 4 Gross and Microscopic, RB	No, (Bill Only)	No

Testing Algorithm

A battery of enzyme histochemical stains will be performed on frozen tissue; additional histochemical stains or immunostains may be performed on frozen tissue; other tests can be performed at an additional charge. The reviewing neuromuscular pathologist will determine the need for additional testing. The patient's provided clinical history, creatine kinase values, and electromyography results are helpful in guiding the additional tests.

For all consultations, ancillary testing necessary to determine a diagnosis is ordered at the discretion of the Mayo Clinic neuromuscular pathologist. An interpretation, which includes an evaluation of the specimen and determination of a diagnosis, will be provided within a formal pathology report.

Frozen tissue sent for consultation: Appropriate additional stains may be performed at an additional charge.

Slides sent for consultation: Special stains and studies performed on the case should be sent with the case for review. In order to determine an accurate diagnosis, some of these stains or studies may be deemed to warrant repeat testing, at an additional charge, at the discretion of the reviewing Mayo Clinic neuromuscular pathologist. The interpreting neuromuscular pathologist may also request frozen tissue to perform additional studies considered necessary for diagnosis.

Note: Testing requested by the referring physician (immunostains, etc) may not be performed if deemed unnecessary by the reviewing Mayo neuromuscular pathologist. Electron microscopic studies **are not performed** on muscle biopsy specimens. For more information see Why Electron Microscopy is Not Performed on Muscle Biopsy Specimens.

For more information see Pathology Consultation Ordering Algorithm.

Special Instructions

- Muscle Biopsy Specimen Preparation Instructions
- Muscle Histochemistry Patient Information
- Pathology Consultation Ordering Algorithm
- Why Electron Microscopy is Not Performed on Muscle Biopsy Specimens

Highlights

Our consultative practice strives to provide the highest quality diagnostic consultative service, balancing optimal patient care with a cost-conscious approach that supports the rapid turnaround time for diagnostic results.

Method Name

Muscle Biopsy Surgical Pathology Consultation and/or Review of Outside Material

NY State Available

Yes

Specimen

Specimen Type

Varies

Ordering Guidance

This test is **not appropriate** for inhalation-transmission diseases such as tuberculosis, *Brucella,* measles, and varicella zoster. This test is also **not appropriate** for suspected Creutzfeldt-Jacobs Disease (CJD).

Additional Testing Requirements

Muscle biopsies from different anatomic sites require separate orders and separate specimen vials.

Shipping Instructions

Transport specimen per instructions in Muscle Biopsy Specimen Preparation Instructions.

Necessary Information

All requisition and supporting information must be submitted in English.

Each of the following items is required:

1. All requisitions must be labeled with:

-Patient name, date of birth, and medical record number

-Name and phone number of the referring pathologist or ordering provider

-Anatomic site and collection date

2. Muscle Histochemistry Patient Information (T361)

Specimen Required

Preferred:

Specimen Type: Frozen muscle biopsy tissue

Supplies: Muscle Biopsy Kit (T541)

Collection Instructions: Prepare and transport specimen per instructions in Muscle Biopsy Specimen Preparation Instructions.

Additional Information:

1. All specimens and additional paperwork must be labeled with:

-Two patient identifiers (first and last name, date of birth, or medical record number)

-Pathology accession/case number

-Anatomic site

2. Contact the Mayo Clinic Muscle Laboratory at 800-533-1710 for special problems to maximize benefit of the muscle biopsy.

Acceptable:

Specimen Type: Stained muscle biopsy slides

Collection Instructions:

1. Submit all stains performed on the case.

2. **All specimens must be labeled with anatomic site**.

Forms

Muscle Histochemistry Patient Information (T361) **is required.**

Specimen Minimum Volume

1.5 cm biopsy

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Ambient		
	Frozen (preferred)		

Clinical & Interpretive

Clinical Information

Muscle diseases are a heterogeneous group of diseases that cause muscle weakness, muscle pain, and elevated creatine kinase. They can be acquired and potentially treatable or inherited. A muscle biopsy allows the diagnosis of a specific muscle disease and is helpful for guiding treatment and genetic testing, as well as investigating relevance of genetic

variants of unknown significance.

The Mayo Clinic Muscle Pathology Laboratory is staffed by neuromuscular neurologists with expertise in the whole spectrum of muscle pathology and complementary clinical competence, and research interest in muscle diseases. This unique set of skills ensures the highest quality of diagnostic muscle pathology in the shortest possible time to the patients, regardless of their geographic location.

A series of 14 stains is routinely performed on each frozen muscle specimen. Client-submitted and in-house stained slides are reviewed in conjunction with the provided clinical history, electromyography, and other laboratory findings. A variety of ancillary studies are available (eg, immunohistochemistry, immunofluorescence) to aid in establishing the diagnosis. These ancillary studies are most efficiently utilized and interpreted in the context of the morphologic features. Each muscle pathology report includes a description of findings and diagnosis, as well as a comment to facilitate integration of pathological findings with other laboratory and clinical data and to guide subsequent testing (eg, serological or genetic tests).

The goal is to provide the highest quality diagnostic service, while balancing optimal patient care with a cost-conscious approach to solve difficult diagnostic problems.

Reference Values

An interpretive report will be provided.

Interpretation

Results are reported in a formal neuromuscular pathology report that includes diagnosis and an interpretive comment, if necessary. The formal pathology report is faxed or sent by mail according to the preference of the referring institution.

Cautions

Be aware that poor freezing and improper handling of the muscle tissue may hinder the neuromuscular pathologist's interpretation of the biopsy. It is crucial to provide a properly handled muscle specimen. In this regard, see directions for Muscle Biopsy Specimen Preparation Instructions. A shipping kit is available, Muscle Biopsy Kit (T541), and is recommended for collecting and shipping muscle specimens. It includes a container for the tissue, collection instructions, the Muscle Histochemistry Patient Information, and a box to ship the specimen.

Clinical Reference

1. Engel AG: The muscle biopsy. In: Engel AG, Franzini-Armstrong C, eds. Myology. 3rd ed. McGraw-Hill; 2004:681-690
2. Liewluck T, Sorenson EJ, Walkiewicz MA, Rumilla KM, Milone M. Autosomal dominant distal myopathy due to a novel ACTA1 mutation. Neuromuscul Disord. 2017;27(8):742-746
3. Engel AG, Redhage KR, Tester DJ, Ackerman MJ, Selcen D. Congenital myopathy associated with the triadin knockout syndrome. Neurology. 2017;88(12):1153-1156
4. Niu Z, Pontifex CS, Berini S, et al: Myopathy with SQSTM1 and TIA1 variants. Clinical and pathological features. Front Neurol. 2018;9:147
5. Nicolau S, Liewluck T, Shen XM, Selcen D, Engel AG, Milone M. A homozygous mutation in GMPPB leads to centronuclear myopathy with combined pre- and postsynaptic defects of neuromuscular transmission. Neuromuscul Disord. 2019;29(8):614-617
6. Nicolau S, Liewluck T, Tracy JA, Laughlin RS, Milone M. Congenital myopathies in the adult neuromuscular clinic: Diagnostic challenges and pitfalls. Neurol Genet. 2019;5(4):e341
7. Liewluck T, Niu Z, Moore SA, Alsharabati M, Milone M. ACTA1-myopathy with prominent finger flexor weakness and

rimmed vacuoles. Neuromuscul Disord. 2019;29(5):388-391
8. Nicolau S, Liewluck T, Elliott JL, Engel AG, Milone M. A novel heterozygous mutation in the C-terminal region of HSPB8 leads to limb-girdle rimmed vacuolar myopathy. Neuromuscul Disord. 2020;30(3):236-240

Performance

Method Description
All requests will be processed as a consultation case first. Special studies will be performed only if deemed to be diagnostically indicated. Histochemical and immunohistochemical stains are only performed if frozen muscle biopsy tissue is provided.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
3 to 14 days; Cases requiring additional material or ancillary testing may require additional time.

Specimen Retention Time
6 months

Performing Laboratory Location
Rochester

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
88342 (if appropriate)
88341 (if appropriate)
88346 (if appropriate)
88350 (if appropriate)

88305 (if appropriate)
88313 (if appropriate)
88319 (if appropriate)
88314 (if appropriate)
88321 (if appropriate)
88323 (if appropriate)
88323-26 (if appropriate)
88325 (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MBX	Muscle Path Consult	60570-9

Result ID	Test Result Name	Result LOINC® Value
601767	Interpretation	59465-5
601769	Participated in the Interpretation	No LOINC Needed
601770	Report electronically signed by	19139-5
601771	Addendum	35265-8
601773	Gross Description	22634-0
601822	Case Number	80398-1
601911	Disclaimer	62364-5
603614	Material Received	81178-6